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**Unit 8 Notes, Part 1: The Basics of Mendelian (Classical) Genetics**

Ms. Ottolini, AP Biology

1. How are traits passed down from parents to offspring? (Note: The information below is taken from your Hardy Weinberg Equilibrium Notes from our Evolution Unit! Isn’t it AWESOME how biology is all interconnected? WOW!!!)
2. Recall the following information from Unit 1 Notes, Part 2 (The Importance of Genetic Variation as Fuel for Natural Selection)?

-“DNA (or deoxyribonucleic acid) is a molecule found in the cells of all living things. The code found in DNA determines the inherited traits found in an organism. An inherited trait is one that can be passed from parents to offspring.”

-“We call the particular alleles a person has for a certain trait his/her genotype for that trait. The genotype determines the actual physical trait, or phenotype.”

- “Within human cells, DNA is organized into 46 chromosomes […] 23 of the 46 chromosomes in one of your cells came from your mother, and the other 23 chromosomes came from your father. Each chromosome from your mother has a complementary chromosome from your father that contains the same types of genes in the same places on the chromosome. We call these pairs of complementary maternal and paternal chromosomes “homologous chromosomes.”

1. Because the chromosomes in your cells are arranged in homologous pairs, you have two alleles for each gene. Therefore, the combination of these two alleles determines your genotype for a particular trait. Typically, one allele form will “cover up” or “mask” the expression of the other allele. We call this allele the dominant allele, and we use a capital letter to represent this allele. When present, this allele determines the physical trait or phenotype that is expressed. We call the allele that is covered up or masked the recessive allele, and we use a lowercase letter to represent this allele. There must be two copies of this allele for the recessive phenotype to be expressed.
2. When there are two copies of the same allele, we call a genotype homozygous. There are two types of homozygous genotype. When there are two dominant alleles, we call this genotype homozygous dominant. When there are two recessive alleles, we call this genotype homozygous recessive. When there is one copy of the dominant allele and one copy of the recessive allele, we call this genotype heterozygous. An example is given below.
3. Let’s say there are two alleles for eye color, brown (B) and blue (b). The possible genotypes and their corresponding phenotypes are given below:

|  |  |  |
| --- | --- | --- |
| Genotype Letters | Genotype Descriptions | Phenotypes |
| BB | Homozygous Dominant | Dominant (Brown Eyes) |
| Bb | Heterozygous | Dominant (Brown Eyes) |
| bb | Homozygous Recessive | Recessive (Blue Eyes) |

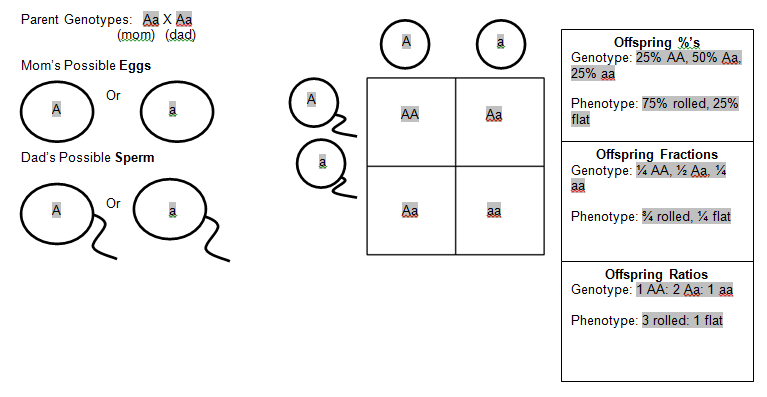
1. Recall the following information from Unit 1 Notes, Part 2?

- “When cells in ovaries (female) or testes (male) divide to create eggs or sperm in the process of meiosis, they must create cells with half the chromosomes of a normal body cell. This way, an egg with 23 chromosomes can meet up with a sperm with 23 chromosomes in a process called fertilization to form a fertilized egg (zygote) with 46 chromosomes. That zygote will then go through normal cell division (mitosis) to create body cells for the fetus with 46 chromosomes each.”

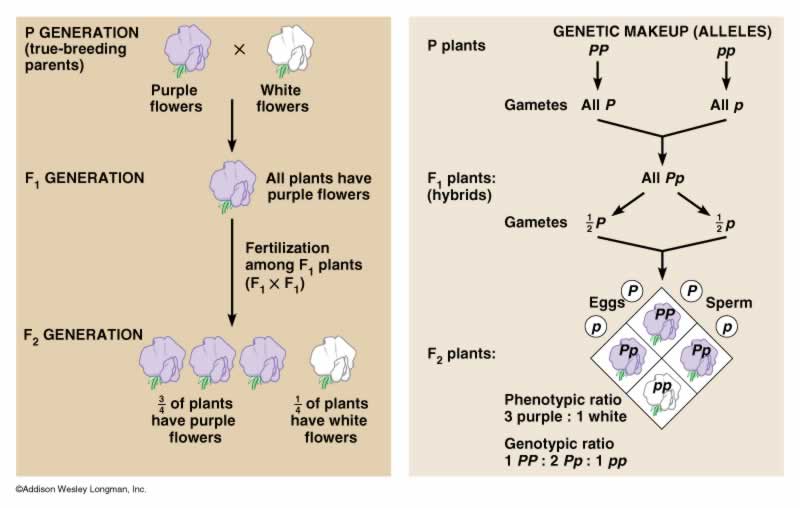
1. Because pairs of homologous chromosomes must divide during meiosis to create eggs and sperm (i.e., gametes) with half the chromosomes found in a normal body cell, each gamete only receives one allele for a particular trait. Therefore, when fertilization occurs, a zygote (and later baby) will receive one allele from each parent. We can use a tool called a Punnett square to predict the possible genotypes for offspring based on possible allele combinations created by the two parental gametes. See the example given below for clarification.

*Note, the axes or sides of the boxes show the alleles found in the gametes from each parent (dad on the left, mom on top). Normally, we do not actually show the eggs and sperm, but I have shown them here for clarification. The inner four boxes show the possible genotypes for offspring (children) from these two parents. Answers are shaded in gray for this example.*

In humans, the ability to roll the tongue is dominant. “**A**” represents the rolled tongue allele, and “**a**” represents the flat tongue allele. Show a cross between a mom and dad that are both **heterozygous** for tongue rolling and find your offspring genotypes and phenotypes in **percentages, fractions, and ratios**.



1. In Gregor Mendel’s famous genetics experiments with pea plants, he conducted genetic “crosses” (breedings) with plants that were purebred for a trait (homozygous). He called these plants the “P” or parent generation. Let’s say one parent is homozygous dominant for purple flowers (PP) and the other parent is homozygous recessive for white flowers (pp).
2. When Mendel allowed these parents (PP x pp) to breed, all the offspring were hybrids (heterozygous). In our example, all the offspring would be heterozygous for purple flowers (Pp). This generation is called the F1 (first filial) generation.
3. When Mendel allowed members of his F1 generation (Pp x Pp) to breed, he saw a 3 : 1 ratio of dominant : recessive phenotypes. In other words, 75% (3/4) of the offspring had the dominant phenotype (purple flowers) and 25% (1/4) of the offspring had the recessive phenotype. (See the image below for details). This “grandbaby generation” is called the F2 (second filial) generation.



1. **How can we calculate the probability of a particular genotype or phenotype in the offspring?**
2. To demonstrate how to do this, let’s look at a practice problem. If green leaves (G) are dominant to yellow leaves (g) in pea plants, what will be the probability of offspring having yellow leaves in a cross between a green, heterozygous parent (Gg) and a yellow parent (gg)? The Punnett square corresponding to this cross is given below

|  |  |  |
| --- | --- | --- |
|  | **G** | **g** |
| **g** | Gg | gg |
| **g** | Gg | gg |

According to the results of the Punnett square, ½ of the offspring have the genotype “gg” and therefore, a yellow phenotype.

1. Now, let’s say we want to determine the probability of an offspring having two specific traits at the same time. Let’s use flower color (purple vs. white) and leaf color (green vs. yellow) as our two traits. If the genotypes of our two parents are PpGg and PpGg (both heterozygous for flower and leaf color), what is the probability that an offspring of these parents would have white flowers AND green leaves?

First, we need to set up a Punnett square for each trait (see below)

|  |  |  |
| --- | --- | --- |
|  | **P** | **p** |
| **P** | PP | Pp |
| **p** | Pp | pp |

*Note: According to the Punnett square above, the probability of the offspring having white flowers is 1/4.*

|  |  |  |
| --- | --- | --- |
|  | **G** | **g** |
| **G** | GG | Gg |
| **g** | Gg | gg |

*Note: According to the Punnett square above, the probability of the offspring having green leaves is 3/4.*

Next, to determine the probability of the offspring having BOTH white flowers AND green leaves (i.e. the probability of two events happening at the same time), we need to multiply their individual probabilities. This is known as the Multiplication Rule of Probabilities. Therefore, the probability of having offspring with white flowers AND green leaves in the problem above is…

1/4 (the probability of having white flowers) x 3/4 (the probability of having green leaves) = 3/16 (the probability of having both white flowers and green leaves)

1. If we used the same problem above but we wanted to determine the probability of offspring having white flowers OR green leaves, we need to add their individual frequencies. When we want to determine the probability of one event OR another event occurring, we need to add their frequencies. This is known as the Addition Rule of Probabilities. For the problem above, the probability of the offspring having white flowers OR green leaves is…

1/4 (the probability of having white flowers) + 3/4 (the probability of having green leaves) = 4/4 = 100% (the probability of having white flowers or green leaves)

1. **Do all traits have a dominant and recessive allele?**
2. No, some traits do not have an allele that is completely dominant over the other, recessive, allele. There are two situations where this is the case…
3. Incomplete dominance occurs when a heterozygous individual has a “blend” of the two phenotypes. In fact, incomplete dominance is often called “blending inheritance.” For example, if an individual receives a tall (T) and short (t) allele, this heterozygous individual (Tt) will be of medium height. As another example, if a flower receives a red (R) and white (r) allele, this heterozygous flower (Rr) will be pink.
4. With our height example, the alleles for tall and short height may be represented using a capital (T) and lowercase (t) letter, even though one allele is not technically dominant over the other. They may also be represented with two different capital letters (ex: T for tall and S for short).
5. Below is an example of a Punnett square problem involving incomplete dominance…

Let’s say that height is a trait controlled by incomplete dominance. We will use “T” to represent the allele for tall height and “S” to represent the allele for short height. If two parents of medium height are crossed, what will be the frequency of tall individuals in the offspring?

|  |  |  |
| --- | --- | --- |
|  | **T** | **S** |
| **T** | TT | TS |
| **S** | TS | SS |

Answer: 1/4 of the offspring will be tall (TT).

1. Codominance, the other “exception to the dominance rule,” occurs when a heterozygous individual displays both traits in their pure form. For example, in cattle, if an individual receives a red hair (R) and white hair (W) allele, this individual (RW) will have some red hairs and some white hairs in its coat. This coloring is called “roan” coloring.
2. Like with incomplete dominance, codominant traits can be represented by a capital and lowercase letter OR two different capital letters.
3. Below is an example of a Punnett square problem involving codominance…

Using the information about coat color codominance in cattle given above, if a red and roan cow are crossed, what will be the frequency of roan individuals in the offspring?

|  |  |  |
| --- | --- | --- |
|  | **R** | **R** |
| **R** | RR | RR |
| **W** | RW | RW |

Answer: 1/2 of the offspring will have a roan coat (RW).

1. **Can we use Punnett squares to predict the inheritance of traits determined by genes located on sex chromosomes?**
2. Humans have 46 chromosomes. There are 22 pairs of autosomes, chromosomes that control normal body traits, and 1 pair of sex chromosomes, chromosomes that control primary and secondary sex characteristics. The image to the right is called a karyotype, and it shows all the chromosomes in a human body cell arranged by homologous pair.
3. Females have two X sex chromosomes (XX) and males have an X chromosome and another, smaller, chromosome called the Y chromosome (XY). We can use Punnett squares to show the inheritance of genes found on the X or Y chromosome (aka sex-linked traits).
4. Even if a trait is controlled by a recessive gene on the X chromosomes, males only need to receive one copy of this gene in order to show the trait (since they only have one X chromosome). Therefore, recessive X-linked traits are more common in males.
5. X-Linked Recessive Traits: Hemophilia is controlled by a RECESSIVE gene on the X chromosome. Therefore, we will use the notation Xh to represent the allele for this condition. We will use the notation XH to represent the normal, dominant allele. Based on this scenario, the following genotypes and phenotypes for males and females will apply…

|  |  |  |
| --- | --- | --- |
| **Genotype** | **Sex** | **Phenotype** |
| XHXH | Female | Normal |
| XHXh | Female | Normal |
| XhXh | Female | Hemophilia |
| XHY | Male | Normal |
| XhY | Male | Hemophilia |

Example Problem: If a mother who is a carrier (heterozygous) for hemophilia marries a man who is normal, what percentage of their male vs. female children will have hemophilia?

|  |  |  |
| --- | --- | --- |
|  | **XH** | **Xh** |
| **XH** | XHXH | XHXh |
| **Y** | XHY | XhY |

Answer: 0% (0/2) of their female children will have hemophilia, and 50% (1/2) of their male children will have hemophilia.

1. X-Linked Dominant Traits: Rett syndrome is controlled by a DOMINANT gene on the X chromosome. Therefore, we will use the notation XR to represent the allele for this condition. We will use the notation Xr to represent the normal, recessive allele. Based on this scenario, the following genotypes and phenotypes for males and females will apply…

|  |  |  |
| --- | --- | --- |
| **Genotype** | **Sex** | **Phenotype** |
| XRXR | Female | Rett |
| XRXr | Female | Rett |
| XrXr | Female | Normal |
| XRY | Male | Rett |
| XrY | Male | Normal |

Example Problem: If a woman who is normal marries a man with Rett syndrome, what percentage of their male vs. female children will have Rett syndrome?

|  |  |  |
| --- | --- | --- |
|  | **Xr** | **Xr** |
| **XR** | XRXr | XRXr |
| **Y** | XrY | XrY |

Answer: 100% (2/2) of their female children will have Rett syndrome, and 0% (0/2) of their male children will have Rett syndrome.

1. **Can we use one Punnett square to show the inheritance of two traits at once?**
2. Up until this point, we have been conducting what are called monohybrid crosses, Punnett squares that show the inheritance of one trait at a time.
3. Now, we will conduct a dihybrid cross, a Punnett square that shows the inheritance of two traits at a time.
4. We can use the multiplication rule of probabilities (see Part B) to determine the probability of an individual inheriting two traits together. However, a dihybrid cross allows us to determine the genotype and phenotype probabilities for all possible combinations of two traits at once. Follow the steps given below to set up and use a Punnett square.
5. Let’s say we are tracking the inheritance of flower and leaf color (see Part B) in plants. Purple flowers (P) are dominant over white flowers (p), and green flowers (G) are dominant over yellow flowers (g). Suppose we want to determine the offspring phenotype frequencies for a cross between parents with the following genotypes:

Parent 1: PpGg

Parent 2: PpGg

To determine the “gametes” that go on the sides and top of your Punnett square, use the “Foil Method” (see below).

*Note: The parents have the same genotype, so they will produce the same gametes. Therefore, I will only show this process once.*

Gamete 1 = F (the FIRST allele for each trait) = PG

Gamete 2 = O (the OUTER allele for each trait) = Pg

Gamete 3 = I (the INNER allele for each trait) = pG

Gamete 4 = L (the LAST allele for each trait) = pg

Once you have determined the gametes for both parents, set up your Punnett square.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
|  | **PG** | **Pg** | **pG** | **pg** |
| **PG** | PPGG | PPGg | PpGG | PpGg |
| **Pg** | PPGg | PPgg | PpGg | Ppgg |
| **pG** | PpGG | PpGg | ppGG | ppGg |
| **pg** | PpGg | Ppgg | ppGg | ppgg |

The genotype frequences are too complicated to calculate, so I will just calculate the phenotype frequencies. First, I need to make sense of my Punnett square. There are four possible phenotypes in my offspring: purple / green (dominant / dominant), purple / yellow (dominant / recessive), white / green (recessive / dominant), and white / yellow (recessive / recessive). I used the following symbols in my Punnett square to denote these phenotypes and came up with the following phenotype frequencies.

|  |  |  |  |
| --- | --- | --- | --- |
| **Phenotype** | **Dominant or Recessive?** | **Symbol** | **Frequency** |
| Purple / Green | Dominant / Dominant |  | 9/16 |
| Purple / Yellow | Dominant / Recessive |  | 3/16 |
| White / Green | Recessive / Dominant |  | 3/16 |
| White / Yellow | Recessive / Recessive |  | 1/16 |

When Mendel did his pea plant experiments with parents who were both heterozygous for two traits, he found that this same phenotype ratio was always present in the offspring…

9 dominant / dominant : 3 dominant / recessive : 3 recessive / dominant : 1 recessive / recessive